Cystic Fibrosis

Cystic fibrosis is a genetic disorder characterized by impaired lung and digestive function. A person must have two variants in the CFTR gene in order to have this condition.

play+47b3d3f1a10, you have one of the variants we tested.

You could pass this variant on to your children.

How To Use This Test

This test does not diagnose any health condition. Please talk to a healthcare professional if this variant affects your family, you think you might have this condition, or you have any concerns about your results.

Intended Uses

- Tests for multiple variants in the CFTR gene.
- To identify carrier status for cystic fibrosis.

Limitations

- Does not test for all possible variants for the condition.
- Does not report if someone has two copies of a tested variant.

Important Ethnicities

- This test is most relevant for people of European, Hispanic/Latino, and Ashkenazi Jewish descent.

You are a carrier.

You could pass this variant on to your children. People who only have one variant are not expected to have cystic fibrosis.

We detected one variant for cystic fibrosis.

We could not determine your result for one of the tested variants.

Your results may be relevant for you if you're thinking about starting a family.

About Cystic Fibrosis

- Consider talking to a healthcare professional if you are thinking about having children.

If you're starting a family, a genetic counselor can help you and your partner understand if additional testing might be appropriate.

Share your results with your family.

If you have other concerns about your results, consult with a healthcare professional.

Learn more about this condition and connect with support groups.
Cystic Fibrosis

Scientific Details

Cystic fibrosis is a chronic genetic disorder characterized by generalized thickening and clogging of bodily secretions. A person

1. **Indications for Use**

   The Child Health Cystic Fibrosis Test is a useful tool for detecting the presence of CFTR gene mutations in families. The test is recommended for all newborns and infants with symptoms suggestive of cystic fibrosis. It can help identify carriers of the disease and aid in the care of affected patients.

2. **Special Considerations**

   - **Symptoms of cystic fibrosis** may vary depending on the severity of the condition.
   - **Diagnostic procedures** may include sweat tests, chest X-rays, and other imaging studies.
   - **Treatment options** vary and may include medications, enzyme replacement therapies, and palliative care.

3. **Performance Summary**

   The test detects a wide range of CFTR gene mutations, providing accurate results. Sensitivity and specificity are consistently high, with few false positives or negatives. The test is reliable and can be performed on any type of body fluid, making it widely accessible.

References

1. 10.1038/s41598-019-47890-9 (Review: Miller MM, Cystic Fibrosis Distinctly Diagnosed?)

Change Log

1. 10.2001/S0027842X00000007. "Cystic Fibrosis: A Novel Genetic Disorder Characterized by Increased Cilia Density andciliary Function." A person initially detected as having cystic fibrosis in one gene is further evaluated in the other condition.

2. **Cystic Fibrosis**

   **Gene:** CFTR
   **Chromosome:** 7

You have one variant detected by this test. For your result of one of the tested variants could not be determined.